IFT80 gene

intraflagellar transport 80

Normal Function

The *IFT80* gene provides instructions for making a protein that is part of a group (complex) called IFT complex B. This complex is found in cell structures known as cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells. IFT complex B is involved in a process called intraflagellar transport (IFT), by which materials are carried within cilia. Specifically, this complex helps transport materials from the base of cilia to the tip.

IFT is essential for the assembly and maintenance of cilia. These cell structures play central roles in many different chemical signaling pathways, including a series of reactions called the Sonic Hedgehog pathway. These pathways are important for the growth and division (proliferation) and maturation (differentiation) of cells. In particular, Sonic Hedgehog appears to be essential for the proliferation and differentiation of cells that ultimately give rise to cartilage and bone.

Health Conditions Related to Genetic Changes

asphyxiating thoracic dystrophy

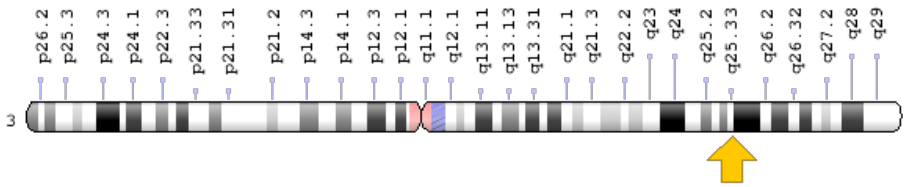
Mutations in the *IFT80* gene were the first genetic changes found to cause asphyxiating thoracic dystrophy, an inherited disorder of bone growth characterized by a small chest, short ribs, and shortened bones in the arms and legs. At least six mutations in the *IFT80* gene have since been associated with this disorder. Most of these mutations change single protein building blocks (amino acids) in the IFT80 protein. IFT complex B made with the altered protein cannot function normally, which disrupts the transport of materials within cilia. Researchers speculate that these changes in IFT alter certain signaling pathways, including the Sonic Hedgehog pathway, which may underlie the abnormalities of bone growth characteristic of asphyxiating thoracic dystrophy.

In some affected individuals, asphyxiating thoracic dystrophy is also associated with abnormalities of the kidneys, liver, retinas, and other tissues. However, when the disorder results from *IFT80* gene mutations, its features are usually limited to problems with bone growth. The reasons for this difference are unknown.

Chromosomal Location

Cytogenetic Location: 3q25.33, which is the long (q) arm of chromosome 3 at position 25.33

Molecular Location: base pairs 160,256,986 to 160,399,532 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ATD2
- IFT80_HUMAN
- intraflagellar transport 80 homolog (Chlamydomonas)
- KIAA1374
- MGC126543
- WD repeat-containing protein 56
- WD repeat domain 56
- WDR56

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Cilia and Flagella: Structure and Movement
  https://www.ncbi.nlm.nih.gov/books/NBK21698/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28IFT80%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- INTRAFLAGELLAR TRANSPORT 80, CHLAMYDOMONAS, HOMOLOG OF
  http://omim.org/entry/611177

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_IFT80.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=IFT80%5Bgene%5D
- HGNC Gene Family: Intraflagellar transport proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/615
- HGNC Gene Family: WD repeat domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/362
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29262
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/57560
- UniProt
  http://www.uniprot.org/uniprot/Q9P2H3

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